

Supporting the Adaptive Responding of Children with Cornelia De Lange Syndrome through Behavioral Interventions: A Mini Review

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Abstract: We reviewed the newest empirical contributions available in the literature on the effectiveness and the suitability of behavioral interventions for supporting the adaptive responding of children with Cornelia de Lange syndrome. According to the eligibility criteria, five studies were retained. Strengths and weaknesses of the selected studies were emphasized. The clinical, psychological and rehabilitative implications of the findings were critically discussed. Finally, the limitations of the current mini review and some useful suggestions for future research and practice were outlined.

Keywords: Cornelia de Lange syndrome, Behavioral Interventions, Quality of Life, Self-Determination, Independence, Adaptive Responding

1. INTRODUCTION

Cornelia de Lange syndrome (CdLs) is a rare genetic disease characterized by extensive motor impairments, growth delays, behavioral disturbances, autistic features, language difficulties, and stereotyped movements [1-2].

The phenotype commonly includes microcephaly, short neck, hirsute ears, highly arched eyebrows, small and widely-spaced teeth, arched palate, downturned mouth corners, and long and thick eyelashes. The range of the related developmental disabilities is usually comprised between the moderate and severe to profound levels [3]. Typically, among the observed behaviors self-injury, hyperactivity, repetitive movements, anxiety, and autistic-like behaviors are included [4-5].

Because children with CdL syndrome constantly rely on caregivers' assistance, a primary rehabilitative goal is the self-determination and the independence of this population towards the environment. To pursue this objective, behavioral interventions, cognitive-behavioral strategies, and assistive technology-based programs (AT) may be profitably considered [6]. Based on learning principles (i.e., causal association between a suitable behavioral response and environmental events), the aforementioned treatments should enhance an active role of a child with CdL syndrome and

prevent his/her isolation and/or passivity. That is, these approaches may promote the participant's social desirability, status, and social image, with positive outcomes on his/her quality of life [7].

Despite the beneficial effects, few evidences on this specific topic are available in the literature [8-10]. For instance, by using CdL and AT as keywords in Scopus database, no documents were found. Switching on CdL and cognitive-behavioral interventions, only two records were carried out [11-12]. Olivet et al. [11] assessed the causal model of significant behaviors in Angelman, CdL, Prader-Willi, and Smith-Magenis syndromes. The authors emphasized the behavioral phenotypes of the analyzed genetic disorders, and described a model of interactions between the physical characteristics and both cognitive and emotional endo-phenotypes, added to the relevant environmental factors. For each syndrome, it was possible to identify pathways from gene to physical phenotype and behavior. The implications of the models for responsive and early interventions were clarified. Cossu, Nava, Leoni, and Piccardi [12] reported a case of mild CdL syndrome, who was exposed to a neuropsychological treatment, which was structured on the specific patient's areas of strengths and weaknesses, in order to fostering the development of adaptive cognitive skills. A multidimensional neuropsychological

approach was recommended. The latter approach should consider the developmental abilities and disabilities of the child as a whole of his/her global reality. Furthermore, only two review papers were found in Scopus if CdL and behavioral interventions were selected as keywords [13-14]. Huisman et al. [13] examined the self-injury behavior (SIB) in CdL among twelve genetic syndromes. They evinced different pattern of SIB, which is closely linked to different gene mutation. Several pathways that led to SIB were identified. Powis and Oliver [14] evaluated and reviewed the prevalence of aggression in genetic syndromes. The results outlined the relevance of phenotype-environment interactions. The future importance of comparison designs, which should examine the assessment and the intervention of aggression was warranted. No reviews assessing the overall effects of specific behavioral, cognitive-behavioral, and AT-based interventions on CdL syndrome were found in Scopus.

Accordingly with the above, the current mini review was a first attempt to emphasize the role of the current literature available on the use of such approach for enhancing adaptive responding and reducing challenging behaviors of children with CdL syndrome. Strengths and weaknesses of the reviewed studies were pointed out. Some useful suggestions for future research and practice within this specific framework were detailed.

2. METHOD

A computerized search was performed in Scopus. A manual search was added as completion. CdL syndrome, AT, behavioral and cognitive-behavioral interventions, quality of life, self-determination, independence, and social validation, indices of happiness, and caregivers burden were merged as keywords. Eligibility criteria were (a) a child and or an adolescent (i.e., range age comprised between 3 and 19 years old) with CdL syndrome, (b) an empirical contribution, either a behavioral or a cognitive-behavioral intervention, and/or an AT-based program, (c) a publication year range included between the 2008 and 2018 (i.e., last decade), and (d) an article written in English language. Accordingly, six papers were selected [15-20], with 210 participants recruited. A brief and concise description was provided for each study, with the rehabilitative objectives, the participants involved, the procedures adopted, and the main results.

3. LITERATURE OVERVIEW

Parisi, Di Filippo, and Roccella [15] described the behavioral phenotype of four children with CdL syndrome and autism spectrum disorders (ASD) with the implemented rehabilitative intervention. Although the symptoms were comparable between ASD and CdL (e.g., repetitive behavior and expressive language delays), the profile and developmental trajectories were quite different between individuals with CdL/ASD and idiopathic ASD. A relevant higher prevalence of self-injuries were closely linked to CdL. Self-injuries were additionally associated with impulsive and repetitive behavior. The suitable rehabilitative interventions implemented were finally described.

Lanovaz, Rapp, and Maciw [16] assessed the effects of a non-contingent reinforcement alone and combined with differential reinforcement of sitting, mouthing, tapping, and appropriate behavior through a single-case experimental design on an individual with CdL and deaf/blindness. Results revealed that non-contingent access to edible items decreased mouthing whereas access to tactile stimuli did not. Combining non-contingent access to tactile items with differential reinforcement reduced mouthing and tapping. Conversely, the appropriate behavior was enhanced.

Moss, Oliver, Nelson, Richards, and Hall [17] examined 130 individuals with CdL, and assessed the ASD profiles. Using the Social Communication Questionnaire, they found a 78.6% of autism in the enrolled participants. However, the profiles were different if compared to typical (i.e., idiopathic) individuals with ASD. A specific association between adaptive behaviors and ASD was additionally identified.

Nakanishi et al. [18] evaluated 49 individuals with the mild to the moderate phenotype of CdL through the Social Communication Questionnaire. The Autism Diagnostic Interview-Revised was additionally administered. A negative correlation between adaptive functioning and ASD was evinced. No differences between gender, age, and genotype were found. A relevant percentage of ASD was emphasized among individuals with CdL. Appropriate interventions were finally recommended.

Richman, Belmont, Kim, Slavin, and Hayner [19] assessed similarities and differences in self-reported stress, patterns of challenging behaviors, and features of autism among 25

participants with CdL (mean age 11.4 years, SD 4.7). A significant percentage of stress was outlined. High levels of challenging behaviors, self-injury, and low levels of adaptive responding could explain such data. Additionally, autism-like behaviors were recorded. Low levels of adaptive responding and communicative skills were pointed out.

Hall, Arron, Sloneem, and Oliver [20] investigated the relationship between self-injury, sleep, and health problems in 54 individuals with CdL, compared with 46 individuals with intellectual disabilities (ID) of mixed etiology, who were similar in terms of ID, mobility, age, and gender through informant-based measures of health/sleep problems and self-injury. Results demonstrated that participants with CdL experienced a higher level of health problems. Conversely, differences between groups with regard to sleep problems were not significantly different. Sleep disorders were not associated with self-injury in CdL participants. Accordingly, CdL individuals were more frequently affected by health problems than other individuals with ID.

4. DISCUSSION

Data of the reviewed studies indicated a specific pattern of physical features in individuals with CdL. Health problems, autism-like behaviors, and self-injury were additionally emphasized. Communication disorders were finally underlined. Behavioral interventions were profitably implemented for promoting adaptive skills and challenging behaviors were consequently reduced. Constructive engagement was enhanced. The findings were consistent with previous empirical contributions [21-24] and suggested the following considerations.

First, behavioral interventions may be useful and helpful for improving adaptive responding of individuals with CdL. Social skills may be increased and self-injury may be prevented, with positive outcomes on their social image, status, social desirability, and quality of life [25-26]. Thus, behavioral interventions based on learning principles (i.e., causal association between adaptive responding and environmental events) may be suitable for teaching purposeful positive behaviors [27].

Second, the challenging behaviors may be redirected toward the adaptive responding. That is, one may argue that by learning new adaptive responding individuals with CdL were no more dealing with the challenging behaviors because constructively engaged and positively occupied.

The pleasant stimuli delivered by the implemented intervention(s). Consequently, individuals with CdL no more exhibited challenging behaviors [28-29].

Third, the systematic comparison between individuals with CdL, ID, and/or other pathologies (e.g., Down syndrome), may allow professionals to designing specific and individualized interventions for persons with CdL. Specific phenotype/genotype trajectories may be described and more specialized treatments may be carried out [30].

5. LIMITATIONS AND FUTURE RESEARCH

Despite the positive outcomes, our mini review presents some limitations. For instance, only six empirical contributions were retained, although few studies were overall available in the literature on CdL. Furthermore, a relatively low number of participants (i.e., two hundred and ten) was considered. Finally, only non-contingent procedure and differential reinforcement of other/alternative behaviors were examined.

In light of the above, new research within this specific framework should consider the following directions. Systematic interventions based on cognitive-behavioral approach and assistive technology-mediated programs should be implemented. New extensions with further empirical investigations should be conducted. An enlargement to new participants with CdL should be assessed. Indices of happiness, and/or positive participation as an outcome measure of the participants' quality of life should be monitored. Preference check assessments should be recorded. Social validation procedures involving external expert raters (e.g., psychologists, caregivers, parents of individuals with developmental disabilities, practitioners, and/or physiotherapists) should be evaluated.

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